CHAIRMAN'S REPORT

YEAR ENDING DECEMBER 31ST 2010

Allan Muir, IPA Chairman

EXTRAORDINARY EXPOSURE

The year started with great excitement about the release of the Hollywood movie, "Extraordinary Measures". After its release in US, and disappointing reviews, its subsequent showing in cinemas around the world was not what we had hoped for. The IPA had prepared press releases to help IPA affiliates capture publicity in their own countries, but sadly the awareness of Pompe disease did not meet our extraordinary expectations. However, the DVD release (and also the showing on some airlines) seems to have received more favourable reviews and will hopefully ensure that there is a constant drip-feed of Pompe Disease awareness around the world.

KEVIN O'DONNELL'S BLOG

At about the same time Kevin, now an advisor to the IPA, completed his history of Pompe ERT development as an online blog: "Pompe Disease - The Real Story" can be found at http://pompestory.blogspot.com/. This is a lengthy document and is essential reading for all those interested in treatments for orphan diseases.

GENZYME CORPORATE DIFFICULTIES

The IPA has watched in dismay as the Genzyme Board of Directors have battled to keep control of the company. When Paul Icahn and other major shareholders threatened to oust Henri Termeer from the chair, the IPA held private teleconferences with Henri to gain a full understanding of the position. We gave our support to Henri because we knew that without his leadership we would never have seen the development of Myozyme, we also had real concerns about future drug development, the financing of Genzyme's charitable programmes (ICAP, InCAP, ATAP) and support to national patient organisations.

Henri won that battle but a new war soon began with the hostile take-over bid for Genzyme from the French company Sanofi-Aventis. This has led to Genzyme's close scrutiny of its finances and the fears we had earlier in the year are still very evident. We have recently heard that a large number of Genzyme staff have lost their jobs and some senior personnel with whom we had built close and trusting relationships have now left the company. Also financial commitments promised to patient organisations (across all orphan diseases) have not been realised; this is a sad time indeed for the company and its stakeholders.

BIOMARIN/ZYSTOR AND AMICUS/GSK

The difficulties at Genzyme have been tempered by the news that BioMarin are taking forward their development of Zystor's GILT technology of an enhanced ERT for Pompe Disease; trials involving Pompe patients are expected to commence in the first quarter of 2011. Amicus Therapeutics is continuing to investigate their Chaperone therapy, but as an adjunct to ERT rather than a mono-therapy. Their research funding for the Pompe programme has reduced to a fraction of its previous levels, but hopefully their new partnership with Glaxo-Smith-Kline will give the project fresh impetus.

POMPE CONNECTIONS

This comprehensive set of brochures was first printed several years ago and we are slowly trying to design updates to reflect the new knowledge about Pompe disease and its treatments since Myozyme was first approved in the US and Europe. There is a constant stream of new information and so managing this process has not been easy, but we are working through the brochures and new issues are posted on our website (www.worldPompe.org). Some updates to the English version were made in July this year; a number of more extensive amendments are planned for the coming months and translations will also be updated as they become available.

COMMUNICATIONS

The secretariat have produced three newsletters this year; these are sent to all IPA affiliated patient groups and are aimed to keep a dialogue going between the board and the membership. A communications subcommittee has recently formed to see whether there is a need to improve the newsletter format and what other measures may be necessary to further involve the membership in our work.

ERT REIMBURSEMENT

A momentous result this year for the US Pompe population, and of course for Genzyme, was the FDA's market approval of Lumizyme (Myozyme to those of us elsewhere in the world). That brought to an end Genzyme's long and expensive compassionate programme (ATAP) for US adults. More importantly it allowed patients, who had been waiting up to 18 months, to benefit from the therapy.

In other countries progress has been mixed with Greece approving Myozyme and yet the Australian government still refusing to reimburse because they do not believe it to have life-saving properties. Genzyme are awaiting the outcome of their recent appeal, a process that has been accompanied by a long (and well received) media campaign by the Australian Pompe Association. Genzyme have resolved to withdraw therapy from adult patients in Australia if Myozyme fails to gain approval and so this is a particularly worrying time for those both on therapy and those waiting to access it.

IPA – ERASMUS- GENZYME (IEG) SURVEY

The original IPA sponsored survey of Pompe adults by the Erasmus Medical Centre has now evolved into the IEG survey that may be completed online or in the original paper form. It is still administrated by the Erasmus MC and this year those enrolled have been asked to complete an update. The survey has also been translated into Spanish and Italian and a survey to study younger participants is being designed.

CONFERENCES, MEETINGS AND THE INTERNET

The AGSD-UK conference this November was a two-day event that attracted speakers and families and professionals from many countries. The IPA took advantage of this and held their AGM and meetings with industry at the same time. There is no doubt that these are vitally important events for families and professionals to meet up and they provide valuable networking opportunities. The IPA is, however, well aware that many families would like to attend such events but are unable to do so due to geographical distance, cost or transportation difficulties. We are currently looking at ways to satisfy the needs of the whole Pompe community and we are encouraged by the example set by the AMDA who have already produced two Webinars (Internet-based seminars): Autophagy by Nina Raben and Gene Therapy by Barry Byrne; both of which were recorded and are available to watch from their

website, www.amda-pompe.org/index.php/main/webinar/. This is a much more controlled method of providing presentations online compared to the recording of speakers at our conferences.

Next year there are plans for Pompe family conferences in the US and Europe and we shall endeavour to maximise the benefit to all families around the globe. Even where internet connections are not high speed or are non-existent.

ENMC Pompe Workshop

I was honoured to be invited by Prof. Ans van der Ploeg and Dr Pascal Laforêt to represent the Pompe patient community at a European NeuroMuscular Centre workshop for leading physicians and researchers. The workshop was structured as a series of presentations by each participant followed by in-depth discussion.

Subjects covered are listed below, a lay report and a full report of the workshop is expected to be posted on the ENMC website very soon (www.enmc.org).

- Outcome of clinical trials with a focus on predicting factors
- Long term clinical effects of enzyme therapy and identification of prognostic factors for infants, children and adults.
- Experiences of physicians treating patients in different European countries and USA.
    - Therapeutic outcome versus natural course
    - good and bad responders
    - potential predicting factors for outcome and relevant adverse events
- Prognostic factors and markers
- New therapies and adaptation of existing therapies
    - chaperones, gene therapy, and others
    - Adaptation of existing therapies: dose changes
    - value of immunotolerization and need for guidelines
- Role of exercise/training
- guidelines for treatment, monitoring and identification of prognostic factors that might be relevant for outcome
- Role of cohort follow-up and industry or country-based registries in the future
- Plans for individual and multicenter clinical trials that the consortium will launch

It was fascinating to hear the professionals struggle with some of the issues that are often discussed by Pompe patients and families and hopefully these discussions will continue for the benefit of the whole Pompe community.

Membership Status

The IPA membership has remained reasonably stable over the year, although we would like to receive more membership fees and more feedback from the many countries we represent. We would always welcome your views on how we can improve the service we provide to the global Pompe community.